ABSTRACT OF THE DISCLOSURES

Disclosed are isolated polynucleotides containing nucleic acid segments encoding human or murine ATP sulfurylase/APS kinase, also known as PAPS synthetase (PAPSS), particularly PAPSS2 and Papss2 proteins. Also disclosed are nucleic acid constructs, including vectors, probes, primers, and primer pairs containing novel *PAPSS2* and *Papss2* gene sequences. A genetically modified vertebrate cell containing a nucleic acid construct of the present invention and a non-human vertebrate comprising the cell are also disclosed. Based on the present *PAPSS2*-specific polynucleotides and nucleic acid constructs, are genetic testing kits and methods for diagnosing spondyloepimetaphyseal dysplasia (SEMD) in a human subject, of identifying a human carrier of an heritable allele associated with SEMD, and of gene therapy or protein therapy for treating a human subject having an osteoarthritic disorder, which is caused or aggravated by deficient enzymatic sulfation activity. Also disclosed is a protein therapy method for treating a human subject having an osteoarthritic disorder caused or aggravated by deficient enzymatic sulfation activity that employs an inventive PAPSS2 fusion protein. Also disclosed are an isolated antibody or antibody fragment that selectively binds a PAPSS2 or Papss2 protein.